LABORATORY BULLETIN

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Laboratory Tests on Newborns for Inborn Errors in Metabolism

NOTE: The number of unsatisfactory specimens received in the laboratory for PKU testing has been increasing. If you experience difficulty in getting enough blood to fill three circles, completely cover one circle rather than try to incompletely fill all circles. One good circle is more desirable than three bad ones.

Revised Codes of Montana 1947, 64-4116 Section 16. Persons in charge of any facility caring for newborn infants and persons responsible for the registration of births shall insure that each infant has a test for phenylketonuria administered under rules adopted by the state board.

By far the most common and practical method for screening large numbers of newborns for phenylketonuria (PKU) is the Guthrie Inhibition Assay. Of 34 states doing the screening tests in their state laboratory in 1970, 32 employed the Guthrie test and two used the Hill fluorometric procedure. During fiscal year '70 our laboratory performed 9,564 Guthrie tests and 2,127 screening tests were performed in three hospitals doing their own tests. Two hospitals use the Guthrie test and one the McCammon and Robbins fluorometric procedure. In our laboratory the test is set up once a week and usually includes 200 specimens. Since inception of the program in 1965, four cases of true PKU have been uncovered in newborns. This yields a ratio of about 1 case for every 14,000 live births. (Nationally this varies from 1:12,000 to 1:15,000.) Last year there were 11,808 live births in Montana so about 117 newborns were missed in the program.

Cost: Our cost in the laboratory for this program last year was $11,477 of which $7,750 came from federal funds through Maternal and Child Health. The cost per test is $1.20 compared with $0.50 in laboratories having a volume of 200,000 tests per year. In figuring costs of laboratory operations the sum of the parts should equal the whole. We begin with our total expenditures and divide them according to the amount spent on each activity which include 16 laboratory tests. Before actual testing is involved, a charge of $0.30 is made against each specimen to include such items as amortization of capital equipment used in reporting, clerical time, copying costs, postage, shipping containers and record keeping. (The last item is particularly important where enforcement is involved as is the case with the PKU law.) The average cost of professional and technical time for testing in our laboratory is now $4.60 per hour and in most instances this accounts for 85 percent or more of the cost of a test. Technical personnel time for PKU costs us $0.80 per test. Our figures cannot be compared with those of non-governmental laboratories because building costs, administrative services received from higher echelons, and allowances for profit and debt service are not included in our budget. The only tests in our laboratory which cost less than $1.00 are the Rubella test (50¢) and serological tests for syphilis (90¢) because with these we are involved in true mass-production operations.

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Screening Tests for Other Inborn Errors of Metabolism: Such tests are available for Galactosemia and Maple Sugar Urine Disease. They can be performed on specimens submitted on the same card as PKU so the $0.30 "handling and reporting" fee would not be repeated. Also, the cost of testing would not be multiplied by three because the same technical skills are used as in PKU testing. The most practical screening test for Maple Sugar Urine Disease is a Guthrie test using another substrate and the Beutler fluorometric procedure has proven most useful for Galactosemia although there is a Guthrie screening test available employing a different organism.

In our opinion, screening tests for these diseases are not justified in Montana. The incidence among newborns is 1:35,000 or lower and both can be recognized by physicians on the basis of characteristic clinical findings in time to institute measures to prevent development of mental retardation. Recently a case of Maple Sugar Urine Disease has been recognized clinically in Montana. Oregon has, since 1965, carried on a program of screening for all three diseases using techniques mentioned above. One case of Maple Sugar Urine Disease was found in 142,219 tests and they encountered one confirmed case of Galactosemia in three years. Cases of inborn errors in metabolism in Oregon are referred to a single hospital center which has a special unit for treatment and study of such patients. In Montana, however, where only one case of PKU may be encountered every two years, it has been more practical to use the referral services offered by the Department of Pediatrics at the University of Colorado Medical School in Denver. New York State screened 141,402 newborns for Galactosemia by a fluorometric method and used one of the blood spots on the PKU card as the specimen. They found four cases; this is about the expected ratio of 1:35,000.

Methods for follow-up and confirmation of positive findings from screening tests include use of paper chromatography, amino acid analyzer and fluorometry. However, none of these methods are practical or suitable for use in mass screening such as one must deal with under PKU laws.